International Congress Series 1288 (2006) 177–179

www.ics-elsevier.com

Common Y-chromosomal STR database for three closely related European populations

Krzysztof Rębała [a,*], Alexei I. Mikulich [b], Iosif S. Tsybovsky [c], Daniela Siváková [d], Zuzana Džupinková [e], Zofia Szczerkowska [a]

[a] *Department of Forensic Medicine, Medical University, Gdansk, Poland*
[b] *Institute for the Study of Arts, Ethnography and Folklore, National Academy of Sciences, Minsk, Belarus*
[c] *Institute of Problems of Criminology, Criminalistics and Forensic Expertise, Minsk, Belarus*
[d] *Department of Anthropology, Comenius University, Bratislava, Slovakia*
[e] *Department of Experimental and Applied Genetics, Institute of Preventive and Clinical Medicine, Medical University, Bratislava, Slovakia*

Abstract. Eighteen Y-chromosomal microsatellite loci (DYS19, DYS388, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS426, DYS437, DYS438, DYS439, DYS460, GATA H4.1, DYS385 a/b, YCAII a/b) were analyzed in 568 males from Belarus, Poland and Slovakia by means of a multiplex (octadecaplex) PCR reaction and capillary electrophoresis in order to compare their usefulness in forensic practice individually and in a haplotype format. Analysis of molecular variance (AMOVA) revealed significant differences between the three populations and excluded one common Y-STR database. Markers responsible for the differences between populations were identified using a chi-square test for homogeneity and locus-by-locus AMOVA. Selection of loci showing homogeneity enabled establishment of a common haplotype database for the studied populations. This approach may become an efficient method for Y-STR databasing for larger ethnogeographic areas as soon as consecutive homogeneity-showing markers are identified.

Keywords: Y-chromosomal short tandem repeat; Belarusian population; Polish population; Slovak population; Database

1. Introduction

A unique inheritance pattern and specificity to males has made the human Y-chromosomal short tandem repeat (Y-STR) markers an excellent tool in male kinship

* Corresponding author. Tel.: +48 58 3491745; fax: +48 58 3410485.
E-mail address: k.rebala@amg.gda.pl (K. Rębała).

0531-5131/
doi:10.1016/j.ics.2005.09.120

analysis, genealogical studies and discrimination of male DNA in male/female stain mixtures [1]. However, extensive analysis of Y-STR polymorphism throughout Europe has shown significant differences in allele and haplotype distribution even between closely related human populations [2,3]. Therefore, establishment of a common Y-STR haplotype frequency database for different European populations appeared to be impossible. Since homogeneity of paternal lineages determined by analysis of minimal haplotypes has been shown between 6 regional populations in Poland [4], the aim of this study was to check if this homogeneity extends to neighbouring closely related Slavic populations of Belarus and Slovakia. Usefulness of 18 Y-chromosomal microsatellites in forensic practice for the three populations was estimated and compared, and a possibility of creation of a common Y-STR haplotype database for forensic purposes was verified.

2. Materials and methods

Eighteen Y-chromosomal microsatellites were genotyped in 360 randomly selected, unrelated males: 196 Belarusians and 164 Slovaks, by means of a multiplex (octadecaplex) PCR reaction and capillary electrophoresis using an ABI Prism 310 Genetic Analyzer, and the results were compared with data already published for 208 unrelated Poles [5]. The loci analyzed included DYS19, DYS388, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS426, DYS437, DYS438, DYS439, DYS460, GATA H4.1, DYS385 a/b, and YCAII a/b. Allele designation was based on comparison with the constructed allelic ladder. In order to check for a marker's potential for resolution of similar haplotypes, contribution of each system to the discrimination capacity was calculated [5]. Analysis of molecular variance (AMOVA) was performed using Arlequin 2.000 software [6]. Markers responsible for the differences between the populations were identified using a chi-square test for homogeneity and locus-by-locus AMOVA.

Table 1
Y-STR genetic diversity and contribution to discrimination capacity in the Belarusian, Polish and Slovak populations

	Genetic diversity			Contribution to discrimination capacity [%]		
	Belarusians ($n=196$)	Poles ($n=208$)	Slovaks ($n=164$)	Belarusians ($n=196$)	Poles ($n=208$)	Slovaks ($n=164$)
DYS19	0.709	0.758	0.741	2.73	5.73	1.29
DYS388	0.461	0.400	0.460	0	0	0.65
DYS389	0.837	0.799	0.806	8.74	9.38	4.52
DYS389I	0.451	0.461	0.431	3.28	2.60	1.94
DYS389II-I	0.689	0.636	0.663	4.92	5.73	1.94
DYS390	0.679	0.654	0.713	1.09	3.13	1.29
DYS391	0.524	0.504	0.519	0	1.56	1.94
DYS392	0.345	0.321	0.394	0.55	0.52	0.65
DYS393	0.348	0.342	0.379	0.55	0	0.65
DYS426	0.498	0.472	0.486	0	0	0
DYS437	0.484	0.457	0.575	0.55	0.52	1.29
DYS438	0.597	0.584	0.657	0	0	0
DYS439	0.703	0.688	0.723	2.19	5.73	2.58
DYS460	0.588	0.563	0.596	2.19	3.13	2.58
GATA H4.1	0.673	0.558	0.635	4.37	2.60	1.94
DYS385	0.829	0.875	0.830	4.92	6.77	3.23
YCAII	0.720	0.638	0.704	1.09	1.56	1.29

3. Results and discussion

The overall haplotype diversity value ranged from 0.9982 in the Polish population to 0.9992 among Belarusians and Slovaks, while discrimination capacity was 93.4%, 92.3%, and 94.5% for Belarusians, Poles and Slovaks, respectively. The most polymorphic system was DYS389 in the Belarusian population, whereas among Poles and Slovaks, the highest gene diversity was found in DYS385. The most valuable marker in discrimination of similar haplotypes was DYS389 while DYS426 and DYS438 did not affect the discrimination power of the multiplex in all three populations (Table 1).

AMOVA revealed significant differences between the populations ($F_{ST}=0.0086$; $P=0.0004$) and excluded possibility of one common Y-STR database. At the significance level equal to 0.05, results obtained by a chi-square test for homogeneity and locus-by-locus AMOVA appeared to be fully consistent, and markers identified as responsible for the population differences were the same. Analysis of haplotypes defined by markers showing homogeneity within the three populations (DYS388, DYS389I, DYS389II-I, DYS393, DYS426, DYS460, YCAII a/b) showed that the whole genetic variation was attributable to the variation within populations ($F_{ST}=-0.0003$; $P=0.49$) and enabled establishment of a common database with haplotype diversity equal to 0.9632 and discrimination capacity equal to 35.4%. For databases combined for pairs of populations, the number of available loci increased (up to 13 loci in case of a Belarusian-Slovak database) and the power of discrimination was higher.

The studied Y-STR loci define very informative haplotypes for population-genetic and forensic investigations. By selecting Y-STR markers showing homogeneity within different populations, we were able to create a common database involving populations that appeared to be genetically different. Such databases eliminate problems of contribution of samples of different ethic background to the gene pool of the general population, which is of importance especially in borderland regions often inhabited by admixed populations. Although elimination of markers including most polymorphic ones led to a significant decrease in haplotype diversity and discrimination capacity, a number of Y-chromosomal microsatellites available for research constantly grows [7] and may enable selection of markers for haplotype databases common for larger ethnogeographic areas, so that discrimination power of such haplotypes may reach a level acceptable in forensic casework.

References

[1] J.M. Butler, Recent developments in Y-short tandem repeat and Y-single nucleotide polymorphism analysis, Forensic Sci. Rev. 15 (2) (2003) 91–111.
[2] L. Roewer, et al., Analysis of molecular variance (AMOVA) of Y-chromosome-specific microsatellites in two closely related human populations, Hum. Mol. Genet. 5 (7) (1996) 1029–1033.
[3] L. Roewer, et al., Signature of recent historical events in the European Y-chromosomal STR haplotype distribution, Hum. Genet. 116 (4) (2005) 279–291.
[4] R. Ploski, et al., Homogeneity and distinctiveness of Polish paternal lineages revealed by Y chromosome microsatellite haplotype analysis, Hum. Genet. 110 (6) (2002) 592–600.
[5] K. Rębała, Z. Szczerkowska, Polish population study on Y chromosome haplotypes defined by 18 STR loci. Int. J. Leg. Med. in press, available online form the Int. J. Leg. Med. 119 (5) (2005) 303–305.
[6] S. Schneider, D. Roessli, L. Excoffier, Arlequin ver. 2.000: A software for population genetics data analysis. Genetics and Biometry Laboratory, University of Geneva, Switzerland, 2000.
[7] M. Kayser, et al., A comprehensive survey of human Y-chromosomal microsatellites, Am. J. Hum. Genet. 74 (6) (2004) 1183–1197.